Opis choroby *

Definicja

Craniosynostosis, Philadelphia type is a form of syndromic craniosynostosis, characterized by sagittal/dolichocephalic head shape with a relatively normal facial appearance and complete soft tissue syndactyly of hand and foot. Transmission is autosomal dominant with variable expression of the hand findings, and incomplete penetrance of the sagittal craniosynostosis. Craniosynostosis, Philadelphia type has been suggested to share the same etiology as syndactyly type 1A.

Dane

Klasyfikacja
Zespół wad wrodzonych

Kod ORPHA
1527

Kod OMIM
185900

Kod ICD10
Q87.0

Kod ICD11
LD24.GY

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*Źródło

orphanet